HLCS gene

holocarboxylase synthetase

Normal Function

The *HLCS* gene provides instructions for making an enzyme called holocarboxylase synthetase. This enzyme is important for the effective use of biotin, a B vitamin found in foods such as liver, egg yolks, and milk. In many of the body's tissues, holocarboxylase synthetase activates enzymes called biotin-dependent carboxylases by attaching biotin to them. These carboxylases are involved in many critical cellular functions, including the production and breakdown of proteins, fats, and carbohydrates.

Holocarboxylase synthetase may also play a role in regulating the activity of genes. In the nucleus, the enzyme likely attaches biotin molecules to histones, which are structural proteins that bind to DNA and give chromosomes their shape. Changing the shape of histones may help determine whether certain genes are turned on or off; however, it is not known how adding biotin affects gene regulation.

Health Conditions Related to Genetic Changes

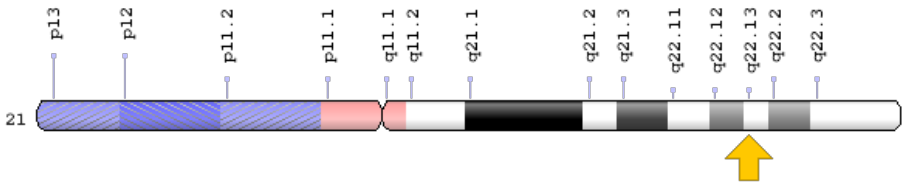
holocarboxylase synthetase deficiency

About 30 mutations in the *HLCS* gene have been identified in people with holocarboxylase synthetase deficiency. Most of these mutations change a single protein building block (amino acid) in the holocarboxylase synthetase enzyme. Many of the known mutations occur in a region of the enzyme that binds to biotin. These genetic changes reduce the enzyme's ability to attach biotin to carboxylases and histones. Without biotin, carboxylases remain inactive and are unable to process proteins, fats, and carbohydrates effectively. A lack of holocarboxylase synthetase activity may also alter the regulation of certain genes that are important for normal development. Researchers believe that these defects in enzyme function underlie the breathing problems, skin rashes, and other characteristic signs and symptoms of holocarboxylase synthetase deficiency.

Chromosomal Location

Cytogenetic Location: 21q22.13, which is the long (q) arm of chromosome 21 at position 22.13

Molecular Location: base pairs 36,750,888 to 36,990,254 on chromosome 21 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- biotin apo-protein ligase
- biotin-protein ligase
- BPL1_HUMAN
- HCS
- holocarboxylase synthetase (biotin-(proprionyl-CoA-carboxylase (ATP-hydrolysing)) ligase)
- holocarboxylase synthetase (biotin-(proprionyl-Coenzyme A-carboxylase (ATP-hydrolysing)) ligase)

Additional Information & Resources

Educational Resources

- Basic Neurochemistry: Molecular, Cellular and Medical Aspects (sixth edition, 1999): Biotin Metabolism
  https://www.ncbi.nlm.nih.gov/books/NBK28072/

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28HLCS%5BTIAB%5D%29+OR+%28holocarboxylase+synthetase%5BTIAB%5D%29%29+OR+%28%28biotin+apo-protein+ligase%5BTIAB%5D%29+OR+%28holocarboxylase+synthetase%5BTIAB%5D%29+OR+%28Biotin-protein+ligase%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D

OMIM

- HOLOCARBOXYLASE SYNTHETASE
  http://omim.org/entry/609018

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_HLCS.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=HLCS%5Bgene%5D
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=4976
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/3141
- UniProt
  http://www.uniprot.org/uniprot/P50747

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